Vulvar extramammary and unilateral Paget’s disease: a case report

Doença de Paget extramamária vulvar e unilateral − Relato de Caso

ABSTRACT

Extramammary Paget’s disease is a rare cutaneous neoplasm typically occurring in the vulva of Caucasian women after menopause. Clinically, the most common symptom is pruritus, when an erythematous, desquamative and eczematous lesion is evidenced. Due to its rarity and nonspecific appearance, the disease can be confused with other dermatological conditions, delaying diagnosis. The present case demonstrates a form of vulvar extramammary unilateral Paget’s disease confirmed by anatomical pathologic and immunohistochemical study. The standard treatment used for the disease in this case was surgical and the patient underwent complete excision of the lesion, with continued monitoring every six months.

Keywords: genital neoplasms, female; vulgar neoplasms; vulvar diseases; paget disease, extramammary.

RESUMO

A doença de Paget extramamária é neoplasia cutânea rara, ocorrendo tipicamente na vulva de mulheres caucasianas na pós-menopausa. Clinicamente, o sintoma mais comum é o prurido, observando-se lesão eritematósa, descamativa e eczematosa. Devido à raridade da doença e a sua aparência inspecífica, pode ser confundida com outras condições dermatológicas, retardando o diagnóstico. O presente caso demonstra uma forma de doença de Paget extramamária vulvar unilateral confirmada por estudo anatomopatológico e imuno-histoquímico. O tratamento-padrão da doença é cirúrgico, e a paciente foi submetida à exérese completa da lesão, continuando em acompanhamento semestral.

Palavras-chave: neoplasias dos genitais femininos; neoplasias vulvares; doenças da vulva; doença de Paget extramamária.

INTRODUCTION

In 1874, Sir James Paget described a breast disease with a very particular histopathology, which was individualized two years later by Butlin. In 1889, Cracker found this very characteristic alteration in a lesion on the scrotum, calling it extramammary Paget’s disease.1

Extramammary Paget’s disease (EMPD) is a rare group of cutaneous neoplasias. It affects both genders, the vulva being the most affected site, followed by the perianal region, perineum, scrotum, and axilae. There are rare reports of the condition in the thighs, buttocks, eyelashes, and external ear.2,3 It typically occurs in patients 60 to 80 years of age, mainly affecting postmenopausal Caucasian women. In Asian populations, however, men are more affected.3

Clinically, the most common symptom is pruritus, and it is evidenced through erythematous, desquamative, and eczematous lesions. It is a slow growing neoplasm in which the appearance of old lesions can be modified by trauma, repeated abrasions, or secondary infection.3 Due to its nonspecific appearance,
it can be mistaken for other dermatological conditions—such as psoriasis, contact dermatitis, squamous cell carcinoma, amelanotic melanoma, and mycosis fungoides—delaying diagnosis.2,3

The standard treatment for EMPD is local surgical exeresis with a 1 cm margin of normal skin, associated with inguinal lymphadenectomy.6 Due to high recurrence rates, which range from 16-50%,7 other therapeutic options, such as Mohs micrographic surgery, photodynamic therapy, radiotherapy, and more recently immunotherapy with imiquimod, have been proposed.8 Regardless of the treatment option, it is crucial to follow up with the patient to detect possible recurrence of the disease as early as possible.

CASE REPORT

A seventy-two-year-old mulatto female patient sought medical attention in February 2010, reporting a vulvar lesion with pruritus, which onset eight years earlier. She reported the use of various topical corticosteroids without improvement. On physical examination, an erythematous, desquamative, crusted, and poorly delimited plaque could be observed in the vulvar and perineal region, on the right hand side of each. There were no palpable lymph nodes in the inguinal region (Figure 1).

The main diagnostic hypotheses were EMPD and chronic eczema. A biopsy of the vulvar region was then carried out, with the histological (Figure 2) and immunohistochemistry (Figures 3-6) studies confirming EMPD. The thorax radiography, the bilateral mammography, and the abdominal ultrasonography showed no abnormalities.

In September 2010, a vulvectomy was performed on the right hand side, with complete excision of the lesion and superficial inguinal lymphadenectomy. No signs of residual tumor were observed in the post-operative examinations. The patient remains in regular follow up every six months, with no signs of recurrence to date.

DISCUSSION

The occurrence of EMPD corresponds to less than 2% of vulvar neoplasias.5 The patient’s epidemiological characteristics are aligned with the literature’s data: female, 72-years-old and postmenopausal. The 8-year clinical history of symptoms confirms the slow progression of the neoplasia, while pointing out the difficulty of diagnosis and appropriate management of the disease.

The present case demonstrated a rare and unilateral form of vulvar EMPD, confirmed through biopsy and followed by histological analysis, which showed numerous foci represented by atypical cells with large, pale cytoplasm containing large nuclei with distinct nucleoli, extending across the mucosal epithelium in a pagetoid pattern. The immunohistochemistry revealed positivity for cytokeratin 7 (CK7), a sensitive marker, however not specific for EMPD. Negativity to that marker is rare, generally occurring in association with malignancy in internal organs.9 Negativity was also observed for cytokeratin 20 (CK20), Melan-A and CDX-2. Positive CK20 is more commonly present in EMPD associated with carcinoma,9 therefore the negativity of this marker in the studied case suggests the absence of other neoplasms. In this context, the expression pattern of cytokeratins provides a clue about the presence or absence of internal malignancy. Melan-A is a melanocyte’s differentiation marker, and its negativity practically excludes the presence of amelanotic melanoma. CDX-2 is found when there is a colorectal tumor, being relevant in the diagnosis of other associated neoplasias.

EMPD generally remains restricted to the epidermis, rarely spreading via the lymphatic system.10 Subjacent malignancy ranges from 12-33% of cases, anatomically correlating to the sites of the lesions and neoplasias, with investigation being fundamental to detect internal malignancy.

Being a rare disease, little is known about the most effective treatment, and the standard approach is surgical excision with anatomopathologic evaluation of frozen margins.4 The
due to the rarity of the case, dermatological knowledge for early diagnosis, attention to other concomitant neoplasia, and the choice of appropriate treatment—whether medical or surgical—are crucial.

REFERENCES


